WHAT IS YOUR DIAGNOSIS ?

5 year old male child, second sibling born to consanguinous parents was brought with the complaints of scaly skin lesions on the soles, feet, knees, elbows and generalised dryness of skin since 5 months of age. He had lost all his teeth at the age of 2 years. His elder sibling, a female aged 10 years was reported to be normal. There was no family history of similar problems.

Examination showed generalized mild dryness of skin. Both soles showed fissuring and scaling. Rough scaly patches were seen on dorsa of feet, knee joints and elbows. Central incisors and canines were absent on both upper and lower jaws. Hair, nails and mucous membranes were normal.

Photographs : 1–3.

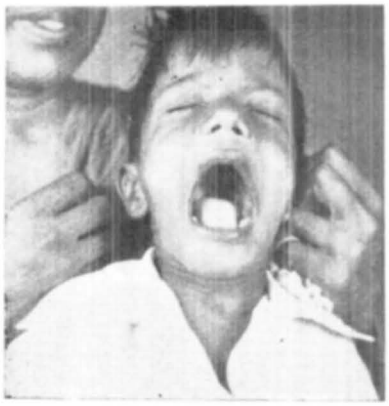

Fig. 1

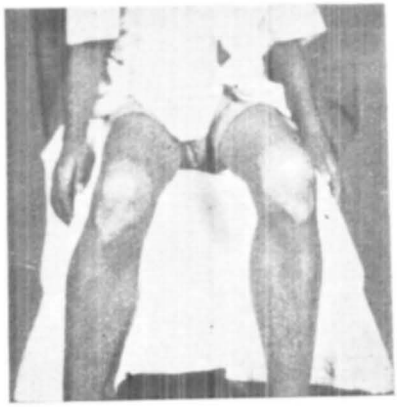

Fig. 2

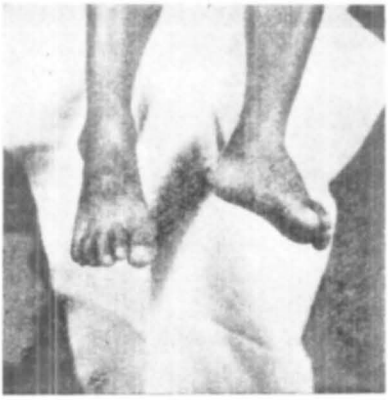

Fig. 3

Differential diagnsois:

1. Papillon–Lefe'vre syndrome
2. X–linked ichthyosis
3. Hidrotic ectodermal dysplasia
4. Mal de meleda

Final Diagnosis : Papillon Lefe'vre syndrome.

The occurrence of dry skin and scaly patches on soles, feet, knees and elbows from early childhood suggested a disorder of keratinisation. The consanguinous parentage suggested a recessively inherited disease. Thus hidrotic ectodermal dysplasia was ruled out. Absence of teeth is not seen in either mal de meleda or X-linked ichyosis.

This child showed plantar hyperkeratosis, circumscribed areas of scaling on elbows, knees and dorsa of feet. Loss of deciduous teeth occurred at the age of 2 years. No evidence of gingival inflammation was present at the time of examination. No other abnormalities were detected. The combination of skin and teeth abnormalities in this recessively inheritied condition suggested the diagnosis of Papillon Lefe'vre syndrome.